

20200903_data_release_notes

Overview of submitted records: 2020

Jan 01, 2020	1,026,969
Feb 01, 2020	1,041,077
Mar 01, 2020	1,055,499
Apr 02, 2020	1,084,731
May 01, 2020	1,136,163
Jun 01, 2020	1,142,645
Jul 01, 2020	1,255,451
Aug 01, 2020	1,302,013
Sep 01, 2020	1,307,377

Overview of changes in the release of September 3, 2020

Content

Brief	Explanation
ClinGen TP53 Variant Curation Expert Panel, ClinGen	ClinGen TP53 Variant Curation Expert Panel, ClinGen submitted 12 novel variant interpretations.
ClinGen Hearing Loss Variant Curation Expert Panel	ClinGen Hearing Loss Variant Curation Expert Panel submitted 11 novel variant interpretations.
Institute of Human Genetics, University of Leipzig Medical Center	Institute of Human Genetics, University of Leipzig Medical Center submitted 1130 novel variant interpretations.
Centre de Biologie Pathologie Génétique, Centre Hospitalier Universitaire de Lille	Centre de Biologie Pathologie Génétique, Centre Hospitalier Universitaire de Lille submitted 1035 novel variant interpretations.
Integrated Genetics /Laboratory Corporation of America	Integrated Genetics/Laboratory Corporation of America submitted 287 novel variant interpretations and updates to 188 records.
Cancer Genomics Group, Japanese Foundation For Cancer Research	Cancer Genomics Group, Japanese Foundation For Cancer Research submitted 298 novel variant interpretations.
Ambry Genetics	Ambry Genetics submitted updates to 236 records.
Dudley Research Group, Pacific Northwest Research Institute	Dudley Research Group, Pacific Northwest Research Institute submitted 224 novel interpretations for variants in the PSAT1 gene.
Centogene AG - the Rare Disease Company	Centogene AG - the Rare Disease Company submitted 206 novel variant interpretations.
Blueprint Genetics	Blueprint Genetics submitted 106 novel variant interpretations.

in this release: changes to variant position, reference and alternate alleles in variant_summary.txt	<p>We updated the way that the variant position and reference and alternate alleles are reported in variant_summary.txt, so that variant_summary.txt is more consistent with ClinVar's VCV XML file.</p> <p>The values for referenceAllele and alternateAllele were reverted to represent right-shifted locations. Columns for vcf_pos, vcf_ref, and vcf_alt were added to represent the left-shifted location, as in the VCF standard. These locations are reported so that they are consistent with the data in the XML files. In each row, either vcf_pos/vcf_ref/vcf_alt (variants in scope for our VCF file) OR start/stop/referenceAllele/alternateAllele are provided (variants not in scope for the VCF file), but not both.</p> <p>Other changes:</p> <ul style="list-style-type: none"> • variants that only have a location on NCBI36 are now included • the Type "undetermined variant" is now reported as "variation" to be consistent with the XML files • RCVAccession is now a unique list of accessions, separated by a pipe. • PhenotypeID has the IDs listed when there are 5 or fewer reported conditions; multiple IDs for one trait are separated by a comma, and multiple traits for one RCV are separated by a semicolon. When there are >5 reported conditions, the number of conditions is reported instead. '-' means that there was an error in calculating the ID. • PhenotypeList has the names listed when there are 5 or fewer reported conditions. When there are >5 reported conditions, the number of conditions is reported instead. • SubmitterCategories now reports 4 for "none" for included variants. • For an included variant, VariationID now reports the Variation ID(s) for haplotypes or genotypes that include the variant, as clinvar:12345. <p>These changes are also documented in the README file on the FTP site:</p> <p>https://ftp.ncbi.nlm.nih.gov/pub/clinvar/README.txt</p> <p>Please contact us as clinvar@ncbi.nlm.nih.gov if you have questions or comments about this change.</p>
coming soon: change in how p. expressions are stored in XML	<p>Soon we will change the way we handle p. expressions that are incomplete because they lack an accession.version, e.g. p. Tyr1863Cys. We will store these descriptions in the XML files as alternate names, rather than as HGVS expressions.</p>

Overview of submitted records: 2019

Jan 01, 2019	759562
Feb 07, 2019	778673
Mar 01, 2019	782638
Apr 01, 2019	787656
May 01, 2019	795045
Jun 01, 2019	811551
Jul 01, 2019	819827
Aug 01, 2019	825177
Sept 01, 2019	881419
Oct 01, 2019	888298
Nov 01, 2019	889968
Dec 01, 2019	893196

Overview of submitted records: 2018

Jan 01, 2018	579543
Feb 01, 2018	582113
Mar 01, 2018	593651
Apr 01, 2018	610005
May 01, 2018	645149
Jun 01, 2018	676018
Jul 01, 2018	676575

Aug 01, 2018	685942
Sep 01, 2018	701880
Oct 01, 2018	708726
Nov 01, 2018	715516
Dec 01, 2018	749203

Overview of submitted records: 2017

Jan 01, 2017	396005
Feb 01, 2017	405182
Mar 01, 2017	406220
Apr 01, 2017	446265
May 01, 2017	482941
Jun 01, 2017	486420
Jul 01, 2017	488658
Aug 01, 2017	492592
Sep 01, 2017	504299
Oct 01, 2017	512373
Nov 01, 2017	517157
Dec 01, 2017	519359

Overview of submitted records: 2016

Jan 01, 2016	172867
Feb 01, 2016	176710
Mar 01, 2016	178032
Apr 01, 2016	180549
May 01, 2016	181155
Jun 01, 2016	192617
Jul 01, 2016	204415
Aug 01, 2016	209842
Sep 01, 2016	210200
Oct 01, 2016	213499
Nov 01, 2016	236420
Dec 01, 2016	240042

Overview of submitted records: 2015

Jan 01, 2015	149013
Feb 01, 2015	156999
Mar 01, 2015	162455

Apr 01, 2015	171408
May 01, 2015	172044
Jun 01, 2015	173236
Jul 01, 2015	184506
Aug 01, 2015	154686
Sep 01, 2015	158580
Oct 01, 2015	160538
Nov 01, 2015	170931
Dec 01, 2015	172006

Overview of submitted records: 2014

Jan 01, 2014	68204
Feb 01, 2014	73492
Mar 01, 2014	83343
Apr 01, 2014	111501
May 01, 2014	112349
Jun 01, 2014	117209
Jul 01, 2014	127132
Aug 01, 2014	127557
Sep 1, 2014	143114
Oct 1, 2014	143601
Nov 1, 2014	144117
Dec 1, 2014	148008

Overview of submitted records: 2013

Apr 05, 2013	30333
May 01, 2013	30386
Jun 01, 2013	39047
Jul 01, 2013	39170
Aug 01, 2013	45901
Sep 01, 2013	50263
Oct 01, 2013	52047
Nov 01, 2013	64750
Dec 01, 2013	64881